PARENT FACT SHEET

DISORDER

S-β Thalassemia (Hb S/Th)

CAUSE

Beta thalassemia is a type of inherited blood disorder that can cause anemia (a low number of red blood cells). It affects a person's ability to produce hemoglobin, the protein in red blood cells that delivers oxygen to all parts of the body.
Signs and symptoms of beta thalassemia are severe in the form of the disorder known as thalassemia major and less severe in the form called thalassemia intermedia. Signs and symptoms of thalassemia major appear in the first 2 years of life. Infants become pale and listless, have a poor appetite, grow slowly, and often develop jaundice (yellowing of the skin). The spleen, liver, and heart may also be enlarged. Adolescents with the severe form may experience delayed puberty. Individuals with thalassemia intermedia may have no symptoms or mild symptoms through childhood and adolescence.

IF NOT TREATED

Most children with thalassemia major appear healthy at birth, but during the first year or two of life they become pale, listless and fussy, and have a poor appetite. They grow slowly and often develop jaundice (yellowing of the skin).

Without treatment, the spleen, liver, and heart soon become greatly enlarged. Bones become thin and brittle; face bones become distorted, and children with thalassemia often look alike. Heart failure and infection are the leading causes of death among children with untreated thalassemia major.

TREATMENT OPTIONS

The use of frequent blood transfusions and antibiotics has improved the outlook for children with thalassemia major. When children with thalassemia major are treated with frequent transfusions (generally every 3 to 4 weeks) aimed at keeping their hemoglobin level near normal, many of the complications of thalassemia can be prevented. This form of treatment, referred to as "hypertransfusion," enhances the child's growth and well-being, and usually prevents heart failure and bone deformities.

IF TREATED

Children with thalassemia major who are treated with frequent blood transfusions and iron chelation live 20 to 30 years or longer. Since intensive chelation treatment was introduced only in the 1960s, continuing studies may show that treated individuals are living even longer.

Thalassemia has been cured using bone marrow transplants. However, this form of treatment is possible only for a small minority of patients who have a suitable bone marrow donor, and the transplant procedure is still risky and can result in death.